Human genomic diversity created by the insertion of Alu retroposons.

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Over 500,000 Alu repeats are dispersed throughout primate genomes in a semi-random manner. Alu elements may be divided into subfamilies of different genetic ages. Three subfamilies (Ya5, Ya8 and Yb8) appear to be of very recent evolutionary origin, having largely expanded within the human genome sometime after the divergence of humans from African apes. We have screened a randomly sheared total human genomic library for members of the Ya5 and Ya8 Alu subfamilies. Over 60 individual loci were subjected to DNA sequence analysis, and oligonucleotide primers complementary to the 5' and 3' flanking unique DNA sequences of each element were used in polymerase chain reaction (PCR) based assays to determine the chromosomal location, phylogenetic distribution, and human genetic variability of each Alu repeat. All of the young Alu elements were restricted to the human genome, and appear to have integrated randomly within the human genome. The polymorphic Alu fossil relics displayed a significant amount of variation in the presence/absence allele frequencies within human population groups from different geographic origins. The combination of over twenty identical by descent polymorphic Alu insertions provides an unprecedented set of nuclear markers for resolving human population relationships.

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